

Genetic Counseling

This report to the American Society of Human Genetics was prepared by an Ad Hoc Committee on Genetic Counseling.* The original report was circulated to all members of the American Society of Human Genetics for comments. The president of the society (V. McKusick) prepared the present modified version based on these comments, and it was subsequently approved by the Ad Hoc Committee on Genetic Counseling and the board of directors of the society.

The Committee on Genetic Counseling of the American Society of Human Genetics was charged with the responsibility of attempting to define the elements of an optimal genetic counseling program. Such definition was felt to be necessary in light of the increasing demand for and availability of genetic counseling services, the widening range of methods applicable to the diagnosis and management of genetic disorders, and the ever expanding number of genetically determined entities that have been delineated. Given the assumption that genetic counseling is part of and has a valid role in health care, a problem of immediate and general concern is the attainment and maintenance of quality of counseling. It is with this problem that the statement which follows deals. While much genetic counseling can be and is provided by nonspecialists in the course of regular medical care, this report will be concerned primarily with counseling by specialized counselors and counseling groups.

Any consideration of counseling must ultimately depend on a definition of the term "genetic counseling." Although many definitions have been proposed, the committee believes that the following one best describes the purposes and scope of genetic counseling.

Genetic counseling is a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family to (1) comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management; (2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives; (3) understand the alternatives for dealing with the risk of recurrence; (4) choose the course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards, and to act in accordance with that decision; and (5) to make the best possible adjustment to the dis-

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order in an affected family member and/or to the risk of recurrence of that disorder.

The emphasis in this definition is on the process of communication. Implicit in it, however, is the concept that knowledge of the correct diagnosis and related medical facts and of the applicable genetic considerations is basic to the entire process. But such knowledge in itself is not sufficient if it cannot be appropriately imparted to the interested person or persons. In addition, knowledge alone will be of only partial benefit to the recipient(s) if assistance is not provided in applying it to the questions with which they are concerned.

The range of problems encompassed in genetic counseling is broad. Included are the wide variety and large number of disorders resulting from single gene abnormalities and chromosomal defects. In addition, there are the conditions believed to be multifactorial in origin—involving multiple genetic factors or a combination of genetic and environmental causes. Prominent in this group are several of the common congenital malformations and, probably, several of the common disorders of adulthood. Many of the situations dealt with in genetic counseling are genetically less well defined. These include multiple malformation syndromes, unclassified mental retardation, recurrent spontaneous abortions, and other instances in which the possibility of a genetic etiology is raised. Other problems include pregnant women exposed to drugs and physical agents, pre-pregnancy parental exposure to drugs and radiation, the potential deleterious effects of consanguinity, the genetic evaluation of children placed for adoption, and premarital counseling.

The tools of the genetic counselor are not in themselves unique, but certain procedural and laboratory methods are heavily relied upon. These include the obtaining of a detailed family history or pedigree, cytogenetic analysis, examination of body fluids for amino acids and other constituents, cell culture and amniocentesis.

Consideration of the range of problems for which counseling is provided and of the tools which must be employed suggests that comprehensive clinical genetic counseling services can best be provided by a team of trained individuals. Since accurate diagnosis of the disorder involved is fundamental to genetic counseling, a physician with clinical experience and specialized training in medical genetics should be a member of the team. (It will always be necessary in special cases to draw on the diagnostic expertise of colleagues in fields such as neurology, ophthalmology, orthopedics, and dentistry.) It is expected that biomedical scientists with Ph.D. degrees and other nonmedical degrees in human or medical genetics will continue to have a vital role in genetic counseling services. Working within the framework of a health team, their role should be the application of the scientific principles and techniques of human genetics to clinically oriented counseling situations. Medical social workers and public health nurses represent professional disciplines which have also been extensively involved with genetic counseling. Members of these two groups tend to function in a similar manner, being prin-

cipally involved in communication between the counseling group and the patient (or patient's family), in the management of social problems, and in obtaining necessary historical and medical information. Another professional group now being trained to function in a like manner is the genetic associates. These individuals receive comprehensive training in human genetics and in communication skills, which prepares them to function as part of a counseling team. A psychiatrist or clinical psychologist has been found to be a useful member of genetic counseling teams.

Training in either basic genetics or medicine alone is not sufficient to qualify an individual as a genetic counselor. A physician or dentist without training or experience specifically in medical genetics or a scientist without training or experience within a specialized medical genetics group is not likely to have the medical, genetic, communication, and laboratory skills necessary for appreciation and handling of the types of problems listed above. While the training program is not precisely defined in either time or content, as a generalization it might reasonably include for medical graduates board eligibility in a branch of clinical medicine, training for which should include *at least* 1 and preferably 2 years within a specialized medical genetics group; for a non-M.D., the Ph.D. degree in genetics or a related field and at least 1 and preferably 2 years of training within a specialized medical genetics group.

With the provision of counseling services to larger segments of the population, the need for appropriately trained nurses, social workers, and/or genetic associates within counseling units will greatly increase. These persons can effectively act as the interface between the geneticists and the patient population, making it possible for the geneticists' time and skills to be most efficiently utilized, while at the same time enhancing the processes of communication and assisting patients and families in dealing with their problems. It is recommended, however, that the extent of their responsibilities for genetic counseling be determined by mutual agreement among the members of the genetic counseling team.

In addition to the professional groups already discussed, genetic counseling units require access to and consultation with physicians who specialize in the various medical disciplines. Specialized laboratory resources are also necessary. While inclusion within the counseling unit of the laboratory facilities and personnel, in particular those involved in cytogenetic and biochemical analyses, is desirable although not essential, the unit under any circumstance must assume responsibility for monitoring the quality of the laboratory work upon which it relies. Therefore, adequate systems of quality control must be applied to these laboratory techniques in the same manner as they are presently applied to more routine types of clinical laboratory analysis.

Quality control should also be applied to the counseling process itself as well as to the laboratory and diagnostic components which back it up. It would be most desirable for each counseling unit to develop or adopt methods to determine how well it is fulfilling the objectives of counseling outlined in the definition and to use such methods on a continuing basis.